

**S2: Young Forensic Scientists Forum – What Shapes
Our Future? Foundations and New Directions**

American Academy of Forensic Sciences
New Orleans, LA (February 14, 2017)



Issues and Challenges with Forensic DNA Analysis

John M. Butler, Ph.D.

NIST Fellow & Special Assistant to the Director for Forensic Science
U.S. National Institute of Standards and Technology



Acknowledgment and Disclaimers

I quote from my recent book entitled “Advanced Topics in Forensic DNA Typing: Interpretation” (Elsevier, 2015). I do not receive any royalties for this book. Completing this book was part of my job at NIST.

Although I chaired the SWGDAM Mixture Committee that produced the 2010 STR Interpretation Guidelines, **I cannot speak for or on behalf of the Scientific Working Group on DNA Analysis Methods.**

I have been fortunate to have had discussions with numerous scientists on interpretation issues including Mike Coble, Bruce Heidebrecht, Robin Cotton, Charlotte Word, Catherine Grgicak, Peter Gill, Ian Evett

...

Points of view are mine and do not necessarily represent the official position or policies of the US Department of Justice or the National Institute of Standards and Technology.

Certain commercial equipment, instruments and materials are identified in order to specify experimental procedures as completely as possible. In no case does such identification imply a recommendation or endorsement by the National Institute of Standards and Technology nor does it imply that any of the materials, instruments or equipment identified are necessarily the best available for the purpose.



Greg Matheson on Forensic Science Philosophy

The CAC News – 2nd Quarter 2012 – p. 6

“Generalist vs. Specialist: a Philosophical Approach”

<http://www.cacnews.org/news/2ndq12.pdf>

- If you want to be a technician, performing tests on requests, then just focus on the policies and procedures of your laboratory. If you want to be a scientist and a professional, learn the policies and procedures, but go much further and learn the philosophy of your profession. **Understand the importance of why things are done** the way they are done, the scientific method, the viewpoint of the critiques, the issues of bias and the importance of ethics.



B.S. Chemistry (1992)

Overview of My Career



UVA Grad Student
(Aug 1992- Aug 1995)

Research Conducted at FBI



**Armed Forces
DNA Identification
Laboratory**

NIST/NRC Postdoc
(Sept 1995- May 1997)

Some Research at AFDIL



Silicon Valley start-up company
doing TOF-MS of DNA

Staff Scientist
(May 1997 – Sept 1999)



**Forensic DNA
Project Leader**

Research Chemist
(Sept 1999 – March 2008)

**Special Assistant to NIST Director for
Forensic Science (April 2013 - present)**

**NIST Fellow &
Group Leader**
(Mar 2008 – Mar 2013)

National Institute of Standards and Technology

- Science agency **part of the U.S. Department of Commerce**
- Started in 1901 as the **National Bureau of Standards**
- Name changed in 1988 to the **National Institute of Standards and Technology (NIST)**
- Forensic science research activities dating back to 1920s
- **Partnership since 2013 with U.S. Department of Justice to create the National Commission on Forensic Science (NCFS) and the Organization of Scientific Area Committees (OSAC)**

- Primary campus in Gaithersburg, Maryland (near Washington, D.C.)
- >3,400 employees and >3,700 associates
- Supplies >1300 reference materials
- Defines official time for the U.S.



DNA reference material

The Best Forensic Scientist You've Never Heard Of

Wilmer Souder and the Early History of Forensic Science
at the National Bureau of Standards



NIST
National Institute of
Standards and Technology
U.S. Department of Commerce



Kristen M. Frederick-Frost, PhD

Robert M. Thompson, BS

John M. Butler, PhD

LW1: **Last Word Society**

American Academy of Forensic Sciences

Las Vegas, NV (February 25, 2016)

NIST FORENSIC SCIENCES



Slides available on the NIST STRBase website:

http://www.cstl.nist.gov/strbase/pub_pres/Souder-AAFS2016-LWS-FINAL.pdf

What Do I Do in My Job at NIST?

- **Write articles** for scientific journals sharing research results or reviewing efforts in forensic science and DNA
 - Have written >150 articles and 5 textbooks so far
- **Prepare presentations** and speak on forensic science and DNA testing to scientists and lawyers and the general public
 - >300 presentations given in >30 states and 25 countries
- **Participate in meetings** influencing forensic science policy and practice
 - Serve as Vice-Chair of the National Commission on Forensic Science
 - Member of the OSAC Biology/DNA Scientific Area Committee
 - Member of the AAFS Standards Board DNA Consensus Body
- **Visit forensic laboratories** to learn of their challenges and to teach them about potential solutions

EVIDENCE TECHNOLOGY MAGAZINE

The magazine dedicated exclusively to the technology of evidence collection, processing, and preservation
Volume 14, Number 2 • Summer 2016



Forensic Science News
from the
National Institute of
Standards and Technology

Recent Activities of the National Commission on Forensic Science

Written by John M. Butler

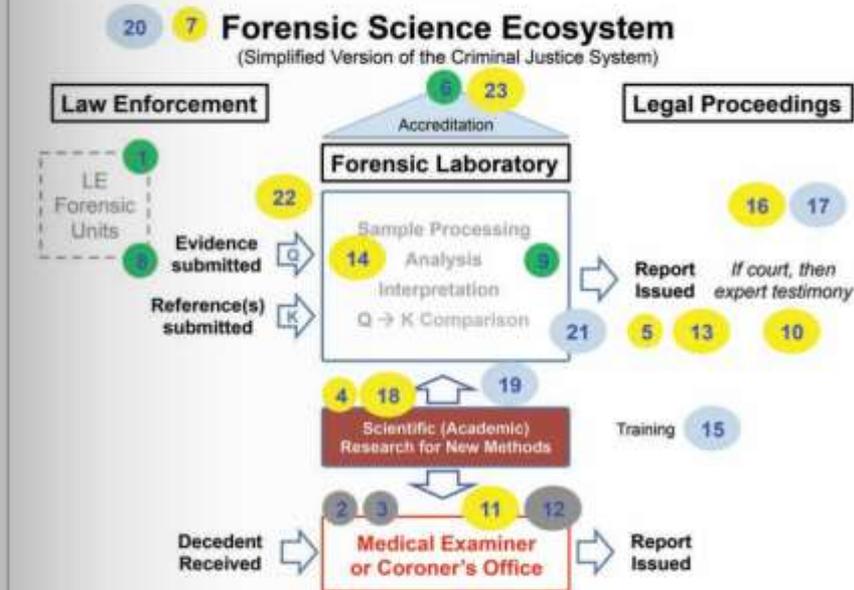
IN FEBRUARY 2013, the U.S. Department of Justice (DOJ) and the National Institute of Standards and Technology (NIST) announced a partnership that included formation of the National Commission on Forensic Science (NCFS) and what is now the Organization of Scientific Area Committees (OSAC). As a Federal Advisory Committee for DOJ, NCFS involves public meetings, public input on draft documents, and an open website sharing meeting materials and final documents. Video recordings of past meetings are available as well. Co-chaired by Deputy Attorney General Sally Yates and NIST Director Willie May, the Commission meets four times a year and involves energetic discussions on a variety of issues.

The accompanying figure is an attempt to show where the 23 NCFS documents thus far approved impact what can be termed the "forensic science ecosystem", which involves law enforcement, forensic laboratories, scientific (academic) research, medical examiner or coroner's offices, and the legal system. For example, NCFS work product #20 is a recommendation regarding a National Code of Professional Responsibility for Forensic Science and Forensic Medicine Service Providers, which received approval at the March 2016 meeting. The number 20 is shaded in light blue because this recommendation is currently under consideration by DOJ.

At the June 20-21, 2016 NCFS meeting, final drafts for seven work products may be introduced for a vote and approval by the Commission. These documents include recommendations regarding pretrial discovery, a request for NIST to perform developmental validation studies, accreditation of digital and multimedia forensic science service providers, and formation of a national disaster call center. Views documents under consideration cover judicial vouching of experts, notice and demand provisions, and validation of forensic science methodology.

The Commission's vision is for all forensic evidence to support the equal and impartial application of justice. The NCFS efforts can be framed into three primary goals: (1) foundational—

NIST CORNER



First 23 NCFS Adopted Work Products

Note: The recommendations in green below and in green on the figure have been responded to by DOJ. Other recommendations are being considered (blue) or are outside the purview of DOJ (gray). Views of the Commission are in yellow. Q = questioned sample; K = known sample

Butler Books on Forensic DNA Typing

2015



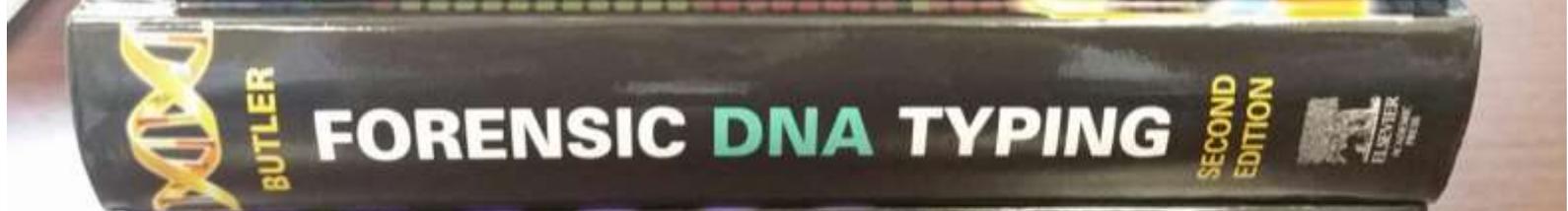
2012



2010



2005



2001



DNA Capabilities to Aid Forensic Investigations

1. The **ability to identify the perpetrator**
2. Weight-of-evidence based on established genetic principles and statistics (Hardy-Weinberg 1908)
3. Established characteristics of genetic inheritance enables close **biological relatives** to be used for reference points using kinship associations
4. Superb **sensitivity** with PCR amplification (opens the possibility for contamination)
5. Well-established **quality assurance measures**
6. New **technology development** aided by genomics

Successful interpretation of DNA (Q-to-K comparison) depends on quality of the crime scene evidence (Q) and availability of suitable reference samples (K)

Forensic DNA Testing in the United States

- We have **~200 public (state and local government) laboratories** performing forensic DNA analysis
 - Two large private companies (Bode Cellmark and Sorenson Forensics) and a few smaller ones perform forensic DNA analysis
- **Over 15 million DNA profiles** in the national DNA database (NDIS: National DNA Index System) run by the FBI Lab
 - Since 1998, the U.S. has included 13 core STR (short tandem repeat) markers; starting in 2017, this number has increased to 20 required STR loci
- **Laboratories have many different protocols** and in some cases, submitting the same sample to two different laboratories could result in two different results
 - Efforts are underway to improve standardization in the field

Critical Challenges Faced Today

- **Success of DNA testing** → significant growth in sample submissions → sample backlogs
 - Laboratory automation and expert system data review
 - Restrictive case acceptance policies to avoid law enforcement investigator ‘swab-athons’ at crime scenes
- **Greater detection sensitivity** → more complex DNA mixtures and low-template DNA with ‘touch’ evidence
 - Probabilistic genotyping to cope with increase in data interpretation uncertainty
 - Use of a complexity threshold to avoid “skating on thin ice”

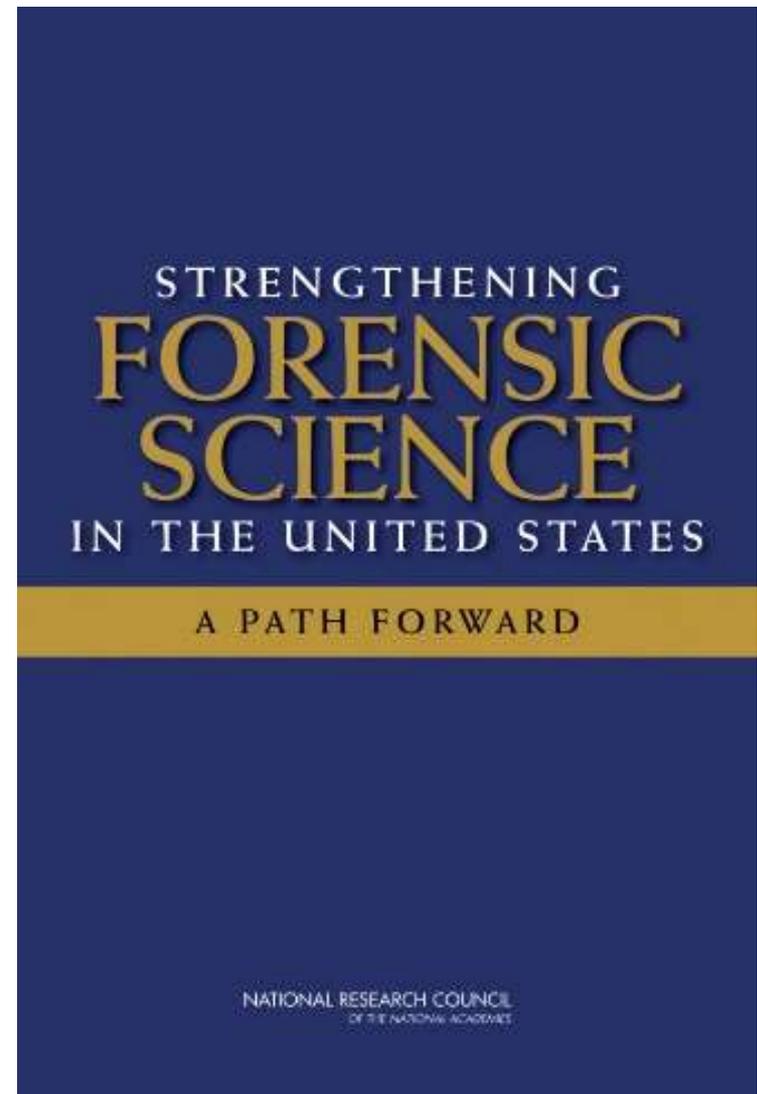
Landmark Report Gives DNA Testing a Pass

Released February 18, 2009

The U.S. National Research Council of the National Academies issued a major report on forensic science in Feb. 2009.

“With the exception of nuclear DNA analysis, no forensic method has been rigorously shown to have the capacity to consistently, and with a high degree of certainty, demonstrate a connection between evidence and a specific individual or source.” (p. 41)

p. 100 mentions limitations with DNA mixtures



Recent Forensic DNA Problems in the News

Washington DC Crime Lab problems with **DNA Mixture Interpretation**

Director of D.C.'s embattled DNA lab
resigns after suspension of testing

Max M. Houck had been the director since the lab opened in 2012. Auditors found major problems there.



Keith L. Alexander and Julie Zauzmer | Local | Apr 30, 2015

April 2015

District could spend nearly \$1 million for outside lab to test
DNA evidence

The District is scrambling to find an alternative after the D.C. lab was ordered to cease DNA testing.

Keith L. Alexander | Crime | Apr 29, 2015

National accreditation board suspends all
DNA testing at D.C. crime lab

Two audits of the District lab found inadequate procedures for DNA testing and poorly trained employees.



Keith L. Alexander | Crime | Apr 28, 2015

The Washington Post

Texas DNA Mixture Case Review

August 2015

<http://www.fsc.texas.gov/texas-dna-mixture-interpretation-case-review>

<http://www.tdcaa.com/journal/changing-state-dna-analysis>

Austin, Texas lab closed in June 2016

Broward County Florida DNA Lab

July 2016

<http://www.browardpalmbeach.com/news/bsc-crime-lab-could-be-mishandling-crucial-dna-evidence-whistleblower-says-7881208>

PCAST Report Comments on Forensic DNA

Released September 20, 2016

- Supports appropriate use of single-source and simple mixture DNA analysis
- **Expresses reservations with complex DNA mixtures** (≥ 3 contributors)

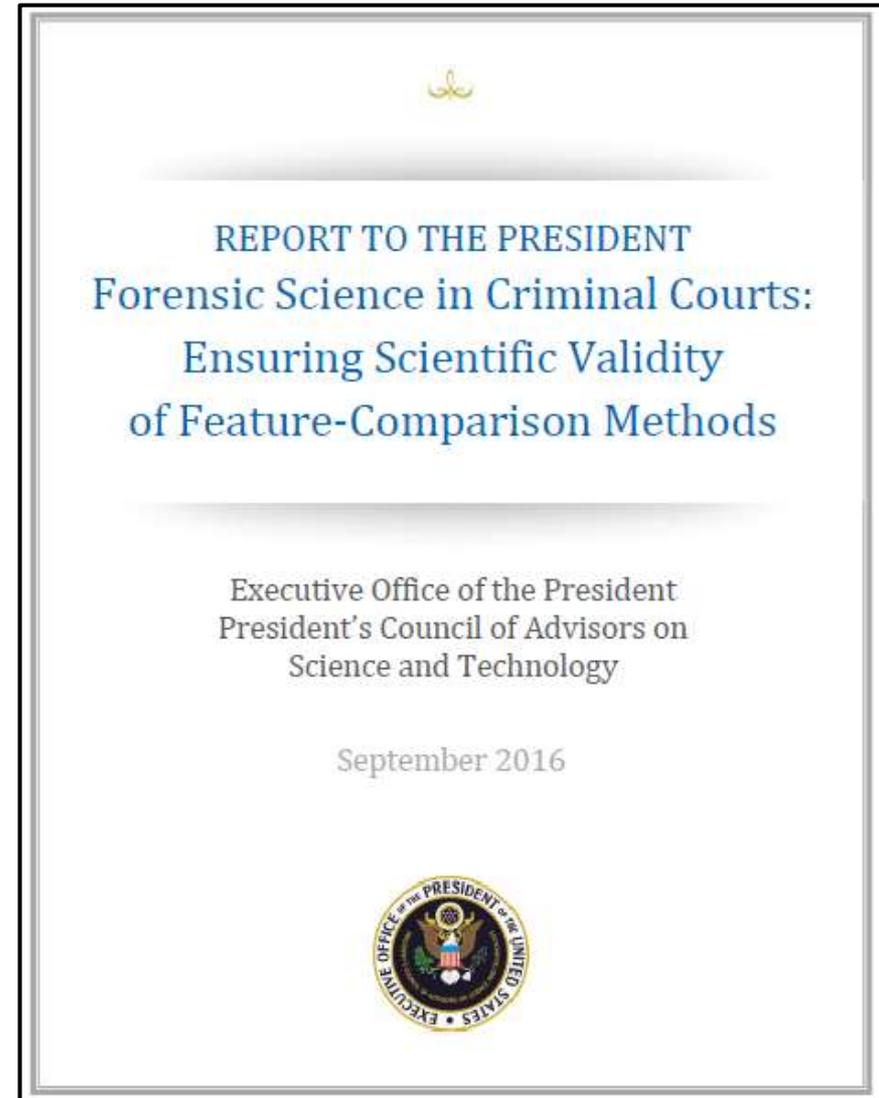
PCAST Co-Chairs



Eric Lander



John Holdren



International conference

The hidden side of DNA profiles. Artifacts, errors and uncertain evidence

Auditorium, Università Cattolica del Sacro Cuore
Rome, 27-28 April, 2012



UNIVERSITÀ
CATTOLICA
del Sacro Cuore



ISFG



SIMLA



GENETISTI FORENSE
ITALIANI



GRUPPO ITALIANO DI
PATOLOGIA FORENSE



David Balding: “Low-template DNA cases are coming to court with limited abilities for sound interpretation. ... There are dangers with LTDNA but we know how to handle and manage them. Unfortunately, proper management is not a universal practice.”



Peter Schneider: “If you cannot explain your evidence to someone that is not from the field (like a judge) – and you need a lot of technical excuses to report something – then the result is not good. You should leave it on your desk and not take it to court. This is a very common sense approach to this problem.”

Thoughts on the Future of Forensic DNA Published in 2015

PHILOSOPHICAL
TRANSACTIONS B

rstb.royalsocietypublishing.org



Opinion piece

CrossMark
click for updates

Cite this article: Butler JM. 2015 The future of forensic DNA analysis. *Phil. Trans. R. Soc. B* **370**: 20140252.

<http://dx.doi.org/10.1098/rstb.2014.0252>

Accepted: 26 February 2015

One contribution of 15 to a discussion meeting issue 'The paradigm shift for UK forensic science'.

The future of forensic DNA analysis

John M. Butler

National Institute of Standards and Technology, Gaithersburg, MD, USA

The author's thoughts and opinions on where the field of forensic DNA testing is headed for the next decade are provided in the context of where the field has come over the past 30 years. Similar to the Olympic motto of 'faster, higher, stronger', forensic DNA protocols can be expected to become more rapid and sensitive and provide stronger investigative potential. New short tandem repeat (STR) loci have expanded the core set of genetic markers used for human identification in Europe and the USA. Rapid DNA testing is on the verge of enabling new applications. Next-generation sequencing has the potential to provide greater depth of coverage for information on STR alleles. Familial DNA searching has expanded capabilities of DNA databases in parts of the world where it is allowed. Challenges and opportunities that will impact the future of forensic DNA are explored including the need for education and training to improve interpretation of complex DNA profiles.

**Addressed Rapid DNA and
Next-Generation Sequencing**

Stages of Forensic DNA Progression

Stages	Time Frame	Description
Exploration	1985 - 1995	Beginnings, different methods tried (RFLP and early PCR)
Stabilization	1995 - 2005	Standardization to STRs, selection of core loci, implementation of Quality Assurance Standards
Growth	2005 - 2015	Rapid growth of DNA databases, extended applications pursued
<i>Sophistication</i>	<i>2015 to 2025 and beyond</i>	<i>Expanding tools available, confronting privacy concerns</i>

Current Trends in Forensic DNA

- ***Faster results:*** Rapid DNA capabilities and new sample-to-answer integrated instruments
- ***Higher sensitivity:*** New assays lowering the limits of detection, which makes interpretation more challenging
- ***Higher information content:*** Next-generation sequencing (NGS) for more markers & STR allele information
- ***Stronger conclusions:*** Mixture interpretation with probabilistic genotyping models

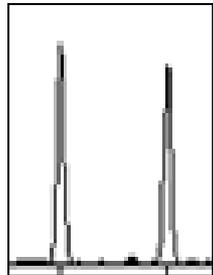
5 Reasons that DNA Results Are Becoming More Challenging to Interpret

1. **More sensitive DNA test results**
2. **More touch evidence samples** that are poor-quality, low-template, complex mixtures
3. **More options exist** for statistical approaches involving probabilistic genotyping software
4. **Many laboratories are not prepared** to cope with complex mixtures
5. **More loci being added** because of the large number of samples in DNA databases

Math Analogy to DNA Evidence

$$2 + 2 = 4$$

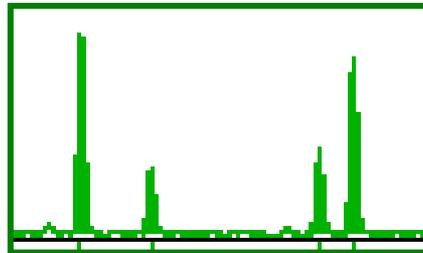
Basic Arithmetic



**Single-Source
DNA Profile**
(DNA databasing)

$$2x^2 + x = 10$$

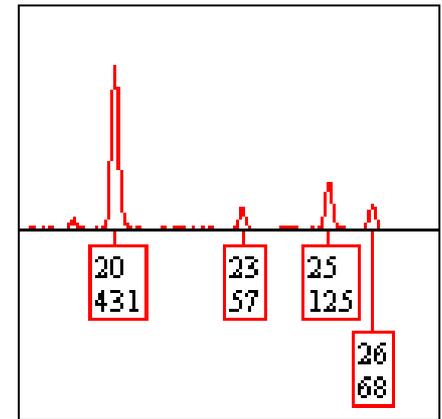
Algebra



Sexual Assault Evidence
(2-person mixture with
high-levels of DNA)

$$\int_{x=0}^{\infty} f(x) dx$$

Calculus



Touch Evidence
(>2-person, low-level,
complex mixtures
perhaps involving
relatives)

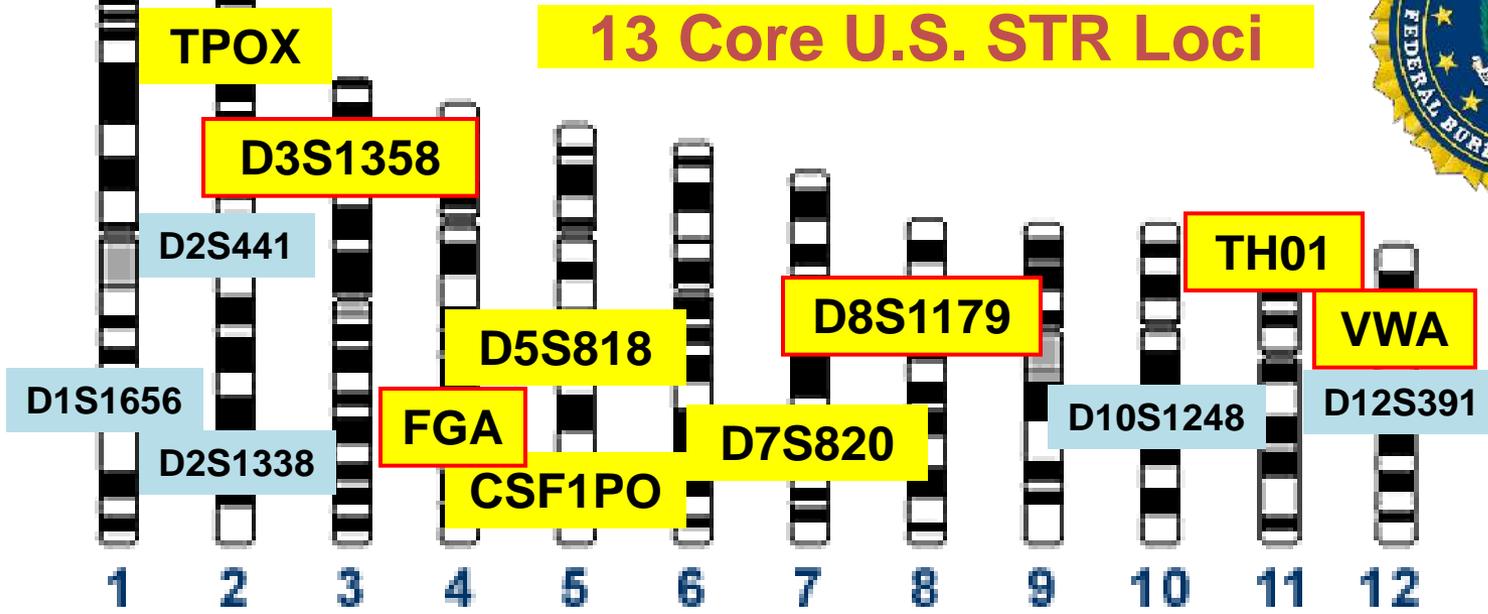
Many laboratories are not prepared to cope with complex mixtures

- Have **appropriate validation studies** been performed to inform proper interpretation protocols? (curriculum & classroom instruction)
- Are **appropriately challenging proficiency tests** being given? (graded homework assignments)
- **Would we want to go into a calculus exam only having studied algebra and having completed homework assignments involving basic arithmetic?**

Position of Forensic STR Markers on Human Chromosomes



Core STR Loci for the United States

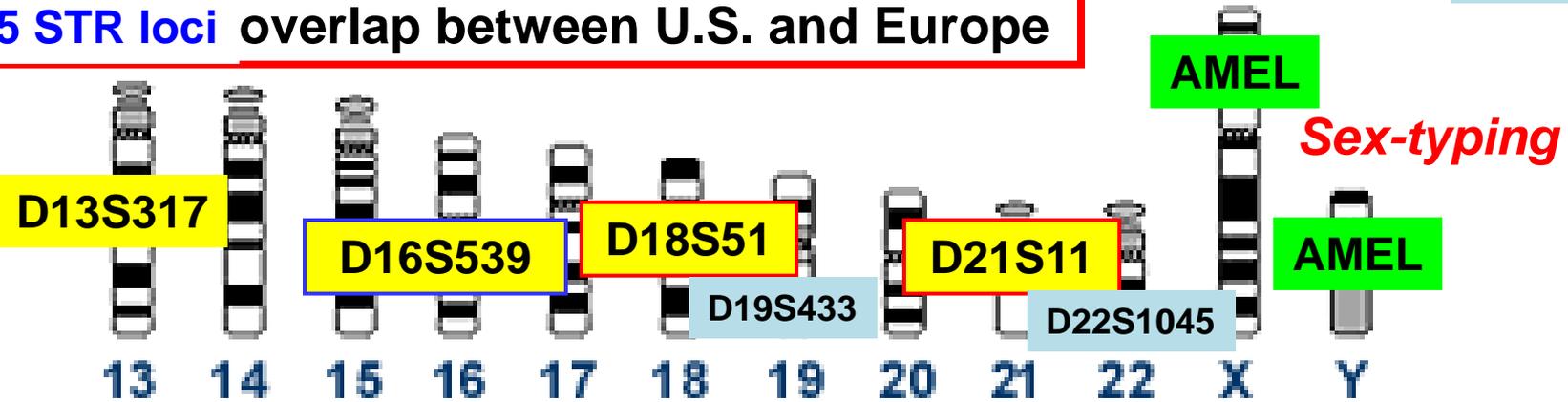


1997
(13 loci)

↓

2017
(20 loci)

15 STR loci overlap between U.S. and Europe



This has required the validation of new DNA testing kits over the past two years

Some Thoughts on Challenges Facing Forensic Science

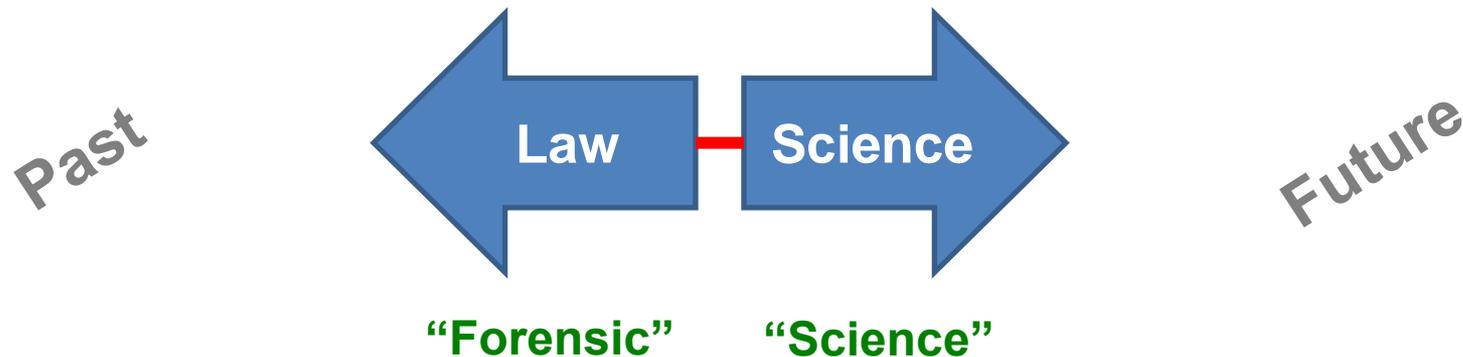
Important Observations

- The National Research Council 2009 (“**NAS Report**”) **called for changes** to strengthen forensic science (with 13 recommendations) but these are not really new issues
- **The criminal justice system**, where forensic science only plays a small part, **is not perfect**; there have been individuals wrongly convicted for a variety of reasons
- Despite a few well-publicized examples (e.g., Annie Dookhan), **forensic scientists** generally want to do a good job and **are trying to do their best**
- **Many forces are at play** to either change things or to maintain the status quo → ***which changes are needed?***

Culture Clash: Science and Law

Tension exists between science and the law:

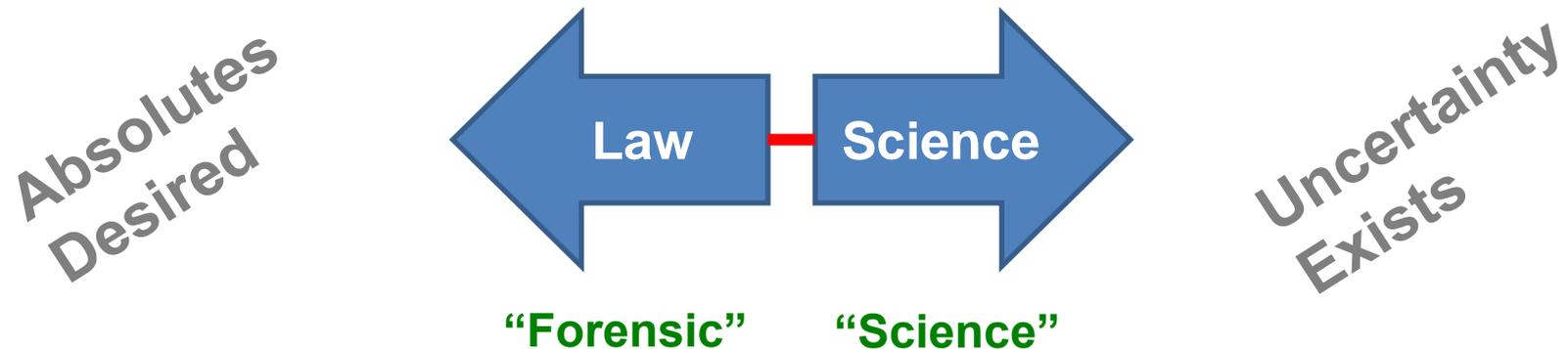
- The legal community **looks to the past**
(precedence is desired)
- The scientific community **looks to the future**
(evolving improvement is desired)



Culture Clash: Science and Law

Tension exists between science and the law:

- The legal community **wants finality and absolutes** (guilty or not-guilty court decisions)
- The scientific community **operates without certainty** (rarely with probabilities of 0 or 1)



Nomenclature Challenges

- We often talk past each other (scientists and lawyers or scientists and scientists) because we do not appreciate a subtle or significant difference in the meaning of a word or phrase
- Examples: “validity” or “validation” can mean something very different to lawyers than to scientists and forensic practitioners
- “A reasonable degree of scientific certainty...”
(a legal crutch that has no scientific meaning)

What Can You Do to Contribute to Solutions in the Future?

Know the literature

Know the question you are trying to answer

Know the limits of what you can do

Know the Literature

- We must do our homework – and read the literature!
- **AAFS 2016 workshop**
 - **Information Does Exist Beyond the First Page of Your Google® Search!:** Tools and Strategies for Forensic Science Literature Searching and Use
 - Search tools and strategies are described
 - Slides available at http://www.cstl.nist.gov/strbase/training/AAFS2016_LiteratureWorkshop.htm

Steps in Forensic DNA Analysis

Gathering the Data

Understanding Results Obtained & Sharing Them

Collection/Storage/
Characterization

Extraction/
Quantitation

Amplification/
Marker Sets

Separation/
Detection

Data

Stats

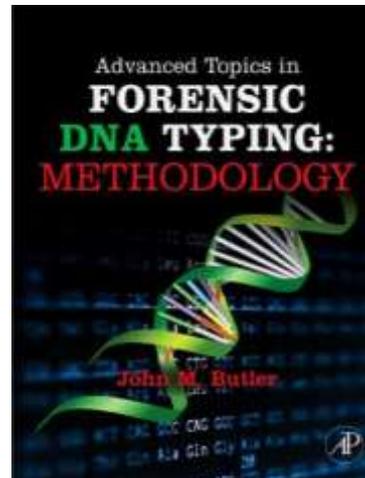
Report

Interpretation

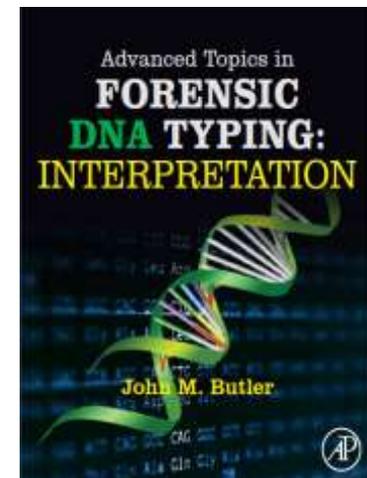
Advanced Topics: Methodology

Advanced Topics: Interpretation

>1300 pages of information with >5000 references cited in these two books



August 2011



October 2014

Know What Question You Are Trying to Answer



David Balding

University of Melbourne
Professor of Mathematics
and Statistics

“...**Focus on the relevant question.** Many misleading statistical approaches [turn] out to be providing valid answers to the wrong questions.”

- David Balding, Interpreting DNA evidence: can probability theory help? In J.L. Gastwirth (ed.) *Statistical Science in the Courtroom* (pp. 51-70) New York: Springer, 2000

Different Calculations Answer Different Questions

Method used	Questions being answered
Profile probability (random match probability, RMP)	What is the rarity of a specific DNA profile given the alleles observed? What is the chance that a particular profile exists in a population based on allele frequencies?
Match probability	Given that a particular profile has been seen (in the crime scene evidence and in the suspect), what is the chance of it occurring again?
Database match probability	How often would a DNA profile match the relevant forensic sample in a database of size N ?



Ian Evett on Interpretation

“The crucial element that the scientist brings to any case is the *interpretation* of those observations. This is the heart of forensic science: it is where the scientist adds value to the process.”

Evett, I.W., et al. (2000). The impact of the principles of evidence interpretation on the structure and content of statements. *Science & Justice*, 40, 233-239.

Know the Limits of What You Can Do

Butler, J.M. (2015) *Advanced Topics in Forensic DNA Typing: Interpretation* (Elsevier Academic Press: San Diego), pp. 159-182

CHAPTER

7

Low-Level DNA and Complex Mixtures

“The limits of each DNA typing procedure should be understood, especially when the DNA sample is small, is a mixture of DNA from multiple sources, or is contaminated with interfering substances.”

NRC I, 1992, p. 8

“For the complex DNA profile, there is no predominant or overarching standard interpretation method.”

Peter Gill (*Gill et al. 2012*, report to the UK Forensic Science Regulator, p. 18)

“The limits of each DNA typing procedure should be understood, especially when the DNA sample is small, is a mixture of DNA from multiple sources...” (NRC I, 1992, p. 8)

*Dans les champs de l'observation le hasard ne favorise
que les esprits préparés
(Inaugural lecture, University of Lille, December 7, 1854)*

Louis Pasteur
(1822-1895)



In the fields of observation **chance favors
only the prepared mind.**

Alexander Hamilton

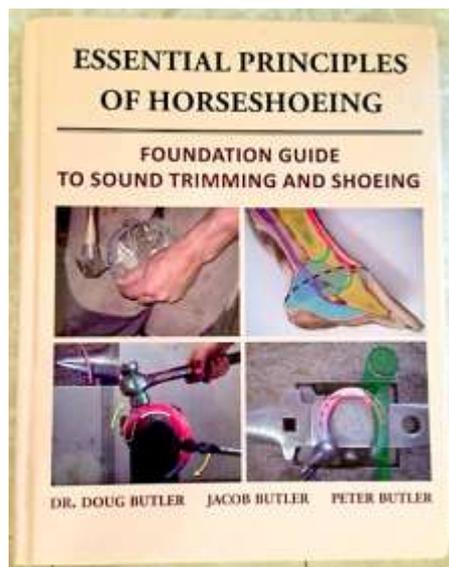
- Men give me some credit for genius, but all of the genius I have lies in this. **When I have a subject in mind, I study it profoundly**, day and night it is before me. I explore it in all its bearings. My mind becomes pervaded with it. The result is what some people call the fruits of genius, whereas **it is in reality the fruits of study and labor** (as quoted in Sterling W. Sill, The Upward Reach, p. 125).

Quote on p. xv, J.M. Butler (2015) *Advanced Topics in Forensic DNA Typing: Interpretation* (Elsevier: San Diego)

Doug Butler Thoughts on Learning

“You never really learn anything until you have to teach it to someone else.”

My father has written a dozen books covering his field of **horseshoeing** and started his own school after teaching at three different universities.



His latest book (2012)



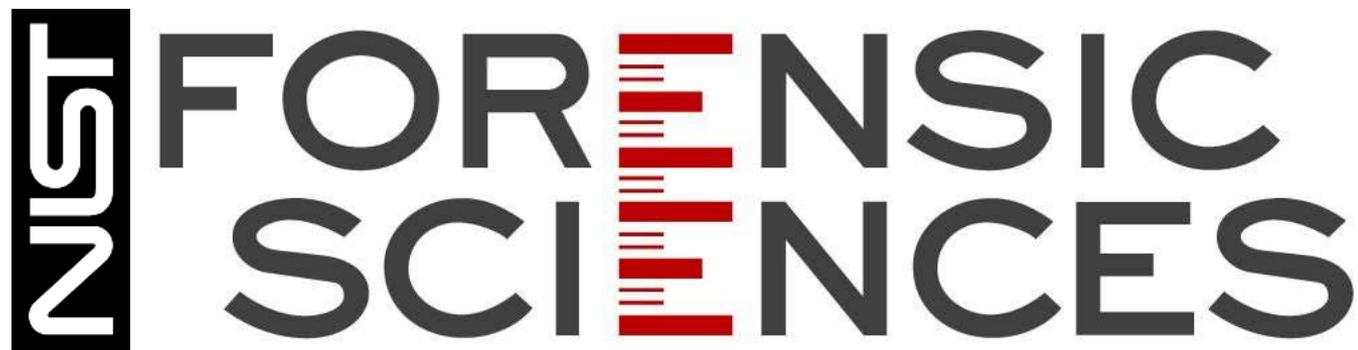
Making horseshoes



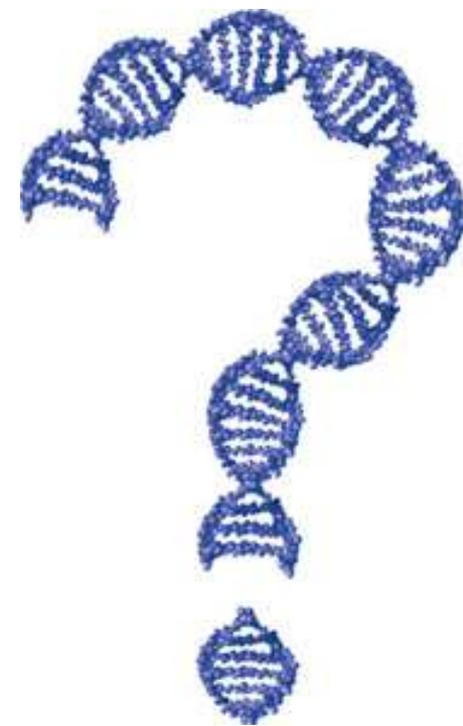
Putting shoes on the horse

National Commission on Forensic Science (NCFS):
www.justice.gov/ncfs

Organization of Scientific Area Committees (OSAC):
www.nist.gov/forensics/osac/index.cfm



www.nist.gov/forensics



+1-301-975-4049

john.butler@nist.gov

International Symposium on Forensic Science Error Management

July 24-28 @NIST, Gaithersburg, MD



Technical Tracks

- Crime Scene
- Death Investigation
- Human Factors
- Legal Factors
- Quality Assurance
- Laboratory Management
- Criminalistics
- Digital Evidence

go.usa.gov/x9yEK

Or search for “NIST 2017 forensic error management”

